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PUBMED 2594783  
REMARK SEQUENCE FROM N.A., AND VARIANT CAIS MET-866.  
REFERENCE 3 (residues 1 to 919)  
AUTHORS Govindan,M.V.  
TITLE Specific region in hormone binding domain is essential for hormone binding and trans-activation by human androgen receptor  
JOURNAL Mol. Endocrinol. 4 (3), 417-427 (1990)  
MEDLINE 90258935  
PUBMED 2342476  
REMARK SEQUENCE FROM N.A.  
REFERENCE 4 (residues 1 to 919)  
AUTHORS Chang,C.S., Kokontis,J. and Liao,S.T.  
TITLE Structural analysis of complementary DNA and amino acid sequences of human and rat androgen receptors  
JOURNAL Proc. Natl. Acad. Sci. U.S.A. 85 (19), 7211-7215 (1988)  
MEDLINE 89017168  
PUBMED 3174628  
REMARK SEQUENCE FROM N.A.  
TISSUE=Prostate  
REFERENCE 5 (residues 1 to 919)  
AUTHORS Tilley,W.D., Marcelli,M., Wilson,J.D. and McPhaul,M.J.  
TITLE Characterization and expression of a cDNA encoding the human androgen receptor  
JOURNAL Proc. Natl. Acad. Sci. U.S.A. 86 (1), 327-331 (1989)  
MEDLINE 89098909  
PUBMED 2911578  
REMARK SEQUENCE FROM N.A.  
TISSUE=Prostate  
REFERENCE 6 (residues 1 to 919)  
AUTHORS Marcelli,M., Tilley,W.D., Wilson,C.M., Griffin,J.E., Wilson,J.D. and McPhaul,M.J.  
TITLE Definition of the human androgen receptor gene structure permits the identification of mutations that cause androgen resistance: premature termination of the receptor protein at amino acid residue 588 causes complete androgen resistance  
JOURNAL Mol. Endocrinol. 4 (8), 1105-1116 (1990)  
MEDLINE 91155943  
PUBMED 2293020  
REMARK SEQUENCE FROM N.A.  
TISSUE=Prostate  
REFERENCE 7 (residues 1 to 919)  
AUTHORS Chang,C.S., Kokontis,J. and Liao,S.T.  
TITLE Molecular cloning of human and rat complementary DNA encoding androgen receptors  
JOURNAL Science 240 (4850), 324-326 (1988)  
MEDLINE 88178111  
PUBMED 3353726  
REMARK SEQUENCE OF 189-919 FROM N.A.  
REFERENCE 8 (residues 1 to 919)  
AUTHORS Trapman,J., Klaassen,P., Kuiper,G.G., van der Korput,J.A., Faber,P.W., van Rooij,H.C., Geurts van Kessel,A., Voorhorst,M.M., Mulder,E. and Brinkmann,A.O.  
TITLE Cloning, structure and expression of a cDNA encoding the human androgen receptor  
JOURNAL Biochem. Biophys. Res. Commun. 153 (1), 241-248 (1988)  
MEDLINE 88240407  
PUBMED 3377788  
REMARK SEQUENCE OF 468-919 FROM N.A.  
REFERENCE 9 (residues 1 to 919)  
AUTHORS Hsiao,P.W., Lin,D.L., Nakao,R. and Chang,C.

TITLE The linkage of Kennedy's neuron disease to ARA24, the first identified androgen receptor polyglutamine region-associated coactivator

JOURNAL J. Biol. Chem. 274 (29), 20229-20234 (1999)

MEDLINE 99329028

PUBMED 10400640

REMARK INTERACTION WITH RAN.

REFERENCE 10 (residues 1 to 919)

AUTHORS Sleddens,H.F.B.M., Oostra,B.A., Brinkmann,A.O. and Trapman,J.

TITLE Trinucleotide repeat polymorphism in the androgen receptor gene (AR)

JOURNAL Nucleic Acids Res. 20, 1427-1427 (1992)

REMARK POLYMORPHISM OF POLY-GLN REGION.

REFERENCE 11 (residues 1 to 919)

AUTHORS Lu,J. and Danielsen,M.

TITLE Direct Submission

JOURNAL Submitted (~FEB-1995)

REMARK POLYMORPHISM OF POLY-GLY REGION.

TISSUE=Blood

REFERENCE 12 (residues 1 to 919)

AUTHORS Giovannucci,E., Stampfer,M.J., Krithivas,K., Brown,M., Dahl,D., Brufsky,A., Talcott,J., Hennekens,C.H. and Kantoff,P.W.

TITLE The CAG repeat within the androgen receptor gene and its relationship to prostate cancer

JOURNAL Proc. Natl. Acad. Sci. U.S.A. 94 (7), 3320-3323 (1997)

MEDLINE 97250535

PUBMED 9096391

REMARK POLYMORPHISM OF POLY-GLN REGION.

REFERENCE 13 (residues 1 to 919)

AUTHORS Giovannucci,E., Stampfer,M.J., Krithivas,K., Brown,M., Dahl,D., Brufsky,A., Talcott,J., Hennekens,C.H. and Kantoff,P.W.

JOURNAL Proc. Natl. Acad. Sci. U.S.A. 94, 8272-8272 (1997)

REMARK ERRATUM.

REFERENCE 14 (residues 1 to 919)

AUTHORS Pinsky,L., Trifiro,M.A., Kaufman,M., Beitel,L.K., Mhatre,A., Kazemi-Esfarjani,P., Sabbaghian,N., Lumbroso,R., Alvarado,C., Vasilious,M. and Gottlieb,B.

TITLE Androgen resistance due to mutation of the androgen receptor

JOURNAL Clin Invest Med 15 (5), 456-472 (1992)

MEDLINE 93092459

PUBMED 1458719

REMARK REVIEW ON VARIANTS.

REFERENCE 15 (residues 1 to 919)

AUTHORS Brown,T.R., Scherer,P.A., Chang,Y.-T., Migeon,C.J., Ghirri,P., Murono,K. and Zhou,Z.

TITLE Molecular genetics of human androgen insensitivity

JOURNAL Eur. J. Pediatr. 152 Suppl. 2, S62-S69 (1993)

REMARK REVIEW ON VARIANTS AIS.

REFERENCE 16 (residues 1 to 919)

AUTHORS Sultan,C., Lumbroso,S., Poujol,N., Belon,C., Boudon,C. and Lobaccaro,J.M.

TITLE Mutations of androgen receptor gene in androgen insensitivity syndromes

JOURNAL J. Steroid Biochem. Mol. Biol. 46 (5), 519-530 (1993)

MEDLINE 94059770

PUBMED 8240973

REMARK REVIEW ON VARIANTS.

REFERENCE 17 (residues 1 to 919)

AUTHORS Patterson,M.N., Hughes,I.A., Gottlieb,B. and Pinsky,L.

TITLE The androgen receptor gene mutations database

JOURNAL Nucleic Acids Res. 22 (17), 3560-3562 (1994)  
MEDLINE 95023089  
PUBMED 7937057  
REMARK REVIEW ON VARIANTS.  
REFERENCE 18 (residues 1 to 919)  
AUTHORS Brinkmann,A.O., Jenster,G., Ris-Stalpers,C., van der Korput,J.A., Bruggenwirth,H.T., Boehmer,A.L. and Trapman,J.  
TITLE Androgen receptor mutations  
JOURNAL J. Steroid Biochem. Mol. Biol. 53 (1-6), 443-448 (1995)  
MEDLINE 95352489  
PUBMED 7626493  
REMARK REVIEW ON VARIANTS.  
REFERENCE 19 (residues 1 to 919)  
AUTHORS Gottlieb,B., Trifiro,M., Lumbroso,R. and Pinsky,L.  
TITLE The androgen receptor gene mutations database  
JOURNAL Nucleic Acids Res. 25 (1), 158-162 (1997)  
MEDLINE 97169385  
PUBMED 9016528  
REMARK REVIEW ON VARIANTS.  
REFERENCE 20 (residues 1 to 919)  
AUTHORS Veldscholte,J., Ris-Stalpers,C., Kuiper,G.G., Jenster,G., Berrevoets,C., Claassen,E., van Rooij,H.C., Trapman,J., Brinkmann,A.O. and Mulder,E.  
TITLE A mutation in the ligand binding domain of the androgen receptor of human LNCaP cells affects steroid binding characteristics and response to anti-androgens  
JOURNAL Biochem. Biophys. Res. Commun. 173 (2), 534-540 (1990)  
MEDLINE 91083633  
PUBMED 2260966  
REMARK VARIANT LNCAP ALA-877.  
REFERENCE 21 (residues 1 to 919)  
AUTHORS Brown,T.R., Lubahn,D.B., Wilson,E.M., French,F.S., Migeon,C.J. and Corden,J.L.  
TITLE Functional characterization of naturally occurring mutant androgen receptors from subjects with complete androgen insensitivity  
JOURNAL Mol. Endocrinol. 4 (12), 1759-1772 (1990)  
MEDLINE 91186983  
PUBMED 2082179  
REMARK VARIANTS CAIS CYS-774; GLN-831 AND MET-866.  
REFERENCE 22 (residues 1 to 919)  
AUTHORS Marcelli,M., Tilley,W.D., Zoppi,S., Griffin,J.E., Wilson,J.D. and McPhaul,M.J.  
TITLE Androgen resistance associated with a mutation of the androgen receptor at amino acid 772 (Arg----Cys) results from a combination of decreased messenger ribonucleic acid levels and impairment of receptor function  
JOURNAL J. Clin. Endocrinol. Metab. 73 (2), 318-325 (1991)  
MEDLINE 91310758  
PUBMED 1856263  
REMARK VARIANT CYS-774.  
REFERENCE 23 (residues 1 to 919)  
AUTHORS Marcelli,M., Zoppi,S., Grino,P.B., Griffin,J.E., Wilson,J.D. and McPhaul,M.J.  
TITLE A mutation in the DNA-binding domain of the androgen receptor gene causes complete testicular feminization in a patient with receptor-positive androgen resistance  
JOURNAL J. Clin. Invest. 87 (3), 1123-1126 (1991)  
MEDLINE 91154385  
PUBMED 1999491  
REMARK VARIANT CAIS PRO-617.

REFERENCE 24 (residues 1 to 919)  
AUTHORS McPhaul,M.J., Marcelli,M., Tilley,W.D., Griffin,J.E.,  
Isidro-Gutierrez,R.F. and Wilson,J.D.  
TITLE Molecular basis of androgen resistance in a family with a  
qualitative abnormality of the androgen receptor and responsive to  
high-dose androgen therapy  
JOURNAL J. Clin. Invest. 87 (4), 1413-1421 (1991)  
MEDLINE 91185626  
PUBMED 2010552  
REMARK VARIANT PAIS CYS-763.  
REFERENCE 25 (residues 1 to 919)  
AUTHORS Ris-Stalpers,C., Trifiro,M.A., Kuiper,G.G.J.M., Jenster,G.,  
Romalo,G., Sai,T., van Rooij,H.C.J., Kaufman,M., Rosenfield,R.L.,  
Liao,S., Schweikert,H.-U., Trapman,J., Pinsky,L. and Brinkmann,A.O.  
TITLE Substitution of aspartic acid-686 by histidine or asparagine in the  
human androgen receptor leads to a functionally inactive protein  
with altered hormone-binding characteristics  
JOURNAL Mol. Endocrinol. 5 (10), 1562-1569 (1991)  
MEDLINE 92131007  
PUBMED 1775137  
REMARK VARIANTS CAIS ASN-695 AND HIS-695, AND SEQUENCE OF 629-723 FROM  
N.A.  
REFERENCE 26 (residues 1 to 919)  
AUTHORS La Spada,A.R., Wilson,E.M., Lubahn,D.B., Harding,A.E. and  
Fischbeck,K.H.  
TITLE Androgen receptor gene mutations in X-linked spinal and bulbar  
muscular atrophy  
JOURNAL Nature 352 (6330), 77-79 (1991)  
MEDLINE 91287825  
PUBMED 2062380  
REMARK VARIANTS SBMA IN POLY-GLN REGION.  
REFERENCE 27 (residues 1 to 919)  
AUTHORS Prior,L., Bordet,S., Trifiro,M.A., Mhatre,A., Kaufman,M.,  
Pinsky,L., Wrogemann,K., Belsham,D.D., Pereira,F., Greenberg,C.R.,  
Trapman,J., Brinkmann,A.O., Chang,C. and Liao,S.  
TITLE Replacement of arginine 773 by cysteine or histidine in the human  
androgen receptor causes complete androgen insensitivity with  
different receptor phenotypes  
JOURNAL Am. J. Hum. Genet. 51 (1), 143-155 (1992)  
MEDLINE 92303560  
PUBMED 1609793  
REMARK VARIANTS CAIS CYS-774 AND HIS-774.  
REFERENCE 28 (residues 1 to 919)  
AUTHORS Saunders,P.T., Padayachi,T., Tincello,D.G., Shalet,S.M. and Wu,F.C.  
TITLE Point mutations detected in the androgen receptor gene of three men  
with partial androgen insensitivity syndrome  
JOURNAL Clin. Endocrinol. (Oxf) 37 (3), 214-220 (1992)  
MEDLINE 93047389  
PUBMED 1424203  
REMARK VARIANTS PAIS LYS-608 AND LEU-866.  
REFERENCE 29 (residues 1 to 919)  
AUTHORS Sweet,C.R., Behzadian,M.A. and McDonough,P.G.  
TITLE A unique point mutation in the androgen receptor gene in a family  
with complete androgen insensitivity syndrome  
JOURNAL Fertil. Steril. 58 (4), 703-707 (1992)  
MEDLINE 93050279  
PUBMED 1426313  
REMARK VARIANT CAIS THR-765.  
REFERENCE 30 (residues 1 to 919)  
AUTHORS Jakubiczka,S., Werder,E.A. and Wieacker,P.

TITLE Point mutation in the steroid-binding domain of the androgen receptor gene in a family with complete androgen insensitivity syndrome (CAIS)  
JOURNAL Hum. Genet. 90 (3), 311-312 (1992)  
MEDLINE 93138625  
PUBMED 1487249  
REMARK VARIANT CAIS VAL-749.  
REFERENCE 31 (residues 1 to 919)  
AUTHORS Batch,J.A., Williams,D.M., Davies,H.R., Brown,B.D., Evans,B.A., Hughes,I.A. and Patterson,M.N.  
TITLE Androgen receptor gene mutations identified by SSCP in fourteen subjects with androgen insensitivity syndrome  
JOURNAL Hum. Mol. Genet. 1 (7), 497-503 (1992)  
MEDLINE 93338440  
PUBMED 1307250  
REMARK VARIANTS CAIS, AND VARIANTS PAIS.  
REFERENCE 32 (residues 1 to 919)  
AUTHORS Nakao,R., Haji,M., Yanase,T., Ogo,A., Takayanagi,R., Katsume,T., Fukumaki,Y. and Nawata,H.  
TITLE A single amino acid substitution (Met786----Val) in the steroid-binding domain of human androgen receptor leads to complete androgen insensitivity syndrome  
JOURNAL J. Clin. Endocrinol. Metab. 74 (5), 1152-1157 (1992)  
MEDLINE 92235226  
PUBMED 1569163  
REMARK VARIANT CAIS VAL-787.  
REFERENCE 33 (residues 1 to 919)  
AUTHORS Wilson,C.M., Griffin,J.E., Wilson,J.D., Marcelli,M., Zoppi,S. and McPhaul,M.J.  
TITLE Immunoreactive androgen receptor expression in subjects with androgen resistance  
JOURNAL J. Clin. Endocrinol. Metab. 75 (6), 1474-1478 (1992)  
MEDLINE 93100381  
PUBMED 1464650  
REMARK VARIANTS CAIS ARG-741 AND CYS-834.  
REFERENCE 34 (residues 1 to 919)  
AUTHORS McPhaul,M.J., Marcelli,M., Zoppi,S., Wilson,C.M., Griffin,J.E. and Wilson,J.D.  
TITLE Mutations in the ligand-binding domain of the androgen receptor gene cluster in two regions of the gene  
JOURNAL J. Clin. Invest. 90 (5), 2097-2101 (1992)  
MEDLINE 93055453  
PUBMED 1430233  
REMARK VARIANTS CAIS, AND VARIANTS PAIS.  
REFERENCE 35 (residues 1 to 919)  
AUTHORS Veldscholte,J., Berrevoets,C.A., Ris-Stalpers,C., Kuiper,G.G., Jenster,G., Trapman,J., Brinkmann,A.O. and Mulder,E.  
TITLE The androgen receptor in LNCaP cells contains a mutation in the ligand binding domain which affects steroid binding characteristics and response to antiandrogens  
JOURNAL J. Steroid Biochem. Mol. Biol. 41 (3-8), 665-669 (1992)  
MEDLINE 92222955  
PUBMED 1562539  
REMARK VARIANT PROSTATE CANCER ALA-877.  
REFERENCE 36 (residues 1 to 919)  
AUTHORS Zoppi,S., Marcelli,M., Deslypere,J.P., Griffin,J.E., Wilson,J.D. and McPhaul,M.J.  
TITLE Amino acid substitutions in the DNA-binding domain of the human androgen receptor are a frequent cause of receptor-binding positive androgen resistance

JOURNAL Mol. Endocrinol. 6 (3), 409-415 (1992)  
MEDLINE 92261595  
PUBMED 1316540  
REMARK VARIANTS CAIS TYR-559 AND ARG-576, AND VARIANTS PAIS GLY-597 AND PRO-617.  
REFERENCE 37 (residues 1 to 919)  
AUTHORS De Bellis,A., Quigley,C.A., Cariello,N.F., el-Awady,M.K., Sar,M., Lane,M.V., Wilson,E.M. and French,F.S.  
TITLE Single base mutations in the human androgen receptor gene causing complete androgen insensitivity: rapid detection by a modified denaturing gradient gel electrophoresis technique  
JOURNAL Mol. Endocrinol. 6 (11), 1909-1920 (1992)  
MEDLINE 93125565  
PUBMED 1480178  
REMARK VARIANTS CAIS SER-705; VAL-749; PHE-759; HIS-774; CYS-855 AND GLY-864.  
REFERENCE 38 (residues 1 to 919)  
AUTHORS Wooster,R., Mangion,J., Eeles,R., Smith,S., Dowsett,M., Averill,D., Barrett-Lee,P., Easton,D.F., Ponder,B.A. and Stratton,M.R.  
TITLE A germline mutation in the androgen receptor gene in two brothers with breast cancer and Reifenstein syndrome  
JOURNAL Nat. Genet. 2 (2), 132-134 (1992)  
MEDLINE 93265137  
PUBMED 1303262  
REMARK VARIANT PAIS/BREAST CANCER GLN-607.  
REFERENCE 39. (residues 1 to 919)  
AUTHORS Newmark,J.R., Hardy,D.O., Tonb,D.C., Carter,B.S., Epstein,J.I., Isaacs,W.B., Brown,T.R. and Barrack,E.R.  
TITLE Androgen receptor gene mutations in human prostate cancer  
JOURNAL Proc. Natl. Acad. Sci. U.S.A. 89 (14), 6319-6323 (1992)  
MEDLINE 92335289  
PUBMED 1631125  
REMARK VARIANT MET-730.  
REFERENCE 40 (residues 1 to 919)  
AUTHORS Macke,J.P., Hu,N., Hu,S., Bailey,M., King,V.L., Brown,T., Hamer,D. and Nathans,J.  
TITLE Sequence variation in the androgen receptor gene is not a common determinant of male sexual orientation  
JOURNAL Am. J. Hum. Genet. 53 (4), 844-852 (1993)  
MEDLINE 94027050  
PUBMED 8213813  
REMARK VARIANTS ARG-205 AND ASP-793.  
REFERENCE 41 (residues 1 to 919)  
AUTHORS Lumbroso,S., Lobaccaro,J.M., Belon,C., Martin,D., Chaussain,J.L. and Sultan,C.  
TITLE A new mutation within the deoxyribonucleic acid-binding domain of the androgen receptor gene in a family with complete androgen insensitivity syndrome  
JOURNAL Fertil. Steril. 60 (5), 814-819 (1993)  
MEDLINE 94039857  
PUBMED 8224266  
REMARK VARIANT CAIS PHE-581.  
REFERENCE 42 (residues 1 to 919)  
AUTHORS Lobaccaro,J.M., Lumbroso,S., Ktari,R., Dumas,R. and Sultan,C.  
TITLE An exonic point mutation creates a MaeIII site in the androgen receptor gene of a family with complete androgen insensitivity syndrome  
JOURNAL Hum. Mol. Genet. 2 (7), 1041-1043 (1993)  
MEDLINE 93372806  
PUBMED 8103398

REMARK VARIANT CAIS VAL-754.  
REFERENCE 43 (residues 1 to 919)  
AUTHORS Lobaccaro,J.M., Lumbroso,S., Belon,C., Galtier-Dereure,F.,  
Bringer,J., Lesimple,T., Namer,M., Cutuli,B.F., Pujol,H. and  
Sultan,C.  
TITLE Androgen receptor gene mutation in male breast cancer  
JOURNAL Hum. Mol. Genet. 2 (11), 1799-1802 (1993)  
MEDLINE 94108428  
PUBMED 8281139  
REMARK VARIANT PAIS/BREAST CANCER LYS-608.  
REFERENCE 44 (residues 1 to 919)  
AUTHORS Adeyemo,O., Kallio,P.J., Palvimo,J.J., Kontula,K. and Janne,O.A.  
TITLE A single-base substitution in exon 6 of the androgen receptor gene  
causing complete androgen insensitivity: the mutated receptor fails  
to transactivate but binds to DNA in vitro  
JOURNAL Hum. Mol. Genet. 2 (11), 1809-1812 (1993)  
MEDLINE 94108430  
PUBMED 8281140  
REMARK VARIANT CAIS ARG-807.  
REFERENCE 45 (residues 1 to 919)  
AUTHORS Nakao,R., Yanase,T., Sakai,Y., Haji,M. and Nawata,H.  
TITLE A single amino acid substitution (gly743 --> val) in the  
steroid-binding domain of the human androgen receptor leads to  
Reifenstein syndrome  
JOURNAL J. Clin. Endocrinol. Metab. 77 (1), 103-107 (1993)  
MEDLINE 93315568  
PUBMED 8325932  
REMARK VARIANT PAIS VAL-743.  
REFERENCE 46 (residues 1 to 919)  
AUTHORS Hiort,O., Huang,Q., Sinnecker,G.H., Sadeghi-Nejad,A., Kruse,K.,  
Wolfe,H.J. and Yandell,D.W.  
TITLE Single strand conformation polymorphism analysis of androgen  
receptor gene mutations in patients with androgen insensitivity  
syndromes: application for diagnosis, genetic counseling, and  
therapy  
JOURNAL J. Clin. Endocrinol. Metab. 77 (1), 262-266 (1993)  
MEDLINE 93315600  
PUBMED 8325950  
REMARK VARIANTS CAIS LYS-681 AND THR-842, AND VARIANTS PAIS HIS-840 AND  
LEU-866.  
REFERENCE 47 (residues 1 to 919)  
AUTHORS Batch,J.A., Evans,B.A., Hughes,I.A. and Patterson,M.N.  
TITLE Mutations of the androgen receptor gene identified in perineal  
hypospadias  
JOURNAL J. Med. Genet. 30 (3), 198-201 (1993)  
MEDLINE 93233131  
PUBMED 8097257  
REMARK VARIANTS PAIS HIS-855 AND MET-869.  
REFERENCE 48 (residues 1 to 919)  
AUTHORS Lobaccaro,J.M., Lumbroso,S., Berta,P., Chaussain,J.L. and Sultan,C.  
TITLE Complete androgen insensitivity syndrome associated with a de novo  
mutation of the androgen receptor gene detected by single strand  
conformation polymorphism  
JOURNAL J. Steroid Biochem. Mol. Biol. 44 (3), 211-216 (1993)  
MEDLINE 93213715  
PUBMED 8096390  
REMARK VARIANT CAIS VAL-743.  
REFERENCE 49 (residues 1 to 919)  
AUTHORS Suzuki,H., Sato,N., Watabe,Y., Masai,M., Seino,S. and Shimazaki,J.  
TITLE Androgen receptor gene mutations in human prostate cancer

JOURNAL J. Steroid Biochem. Mol. Biol. 46 (6), 759-765 (1993)  
MEDLINE 94100129  
PUBMED 8274409  
REMARK VARIANTS PROSTATE CANCER HIS-701 AND ALA-877.  
REFERENCE 50 (residues 1 to 919)  
AUTHORS Kazemi-Esfarjani,P., Beitel,L.K., Trifiro,M., Kaufman,M., Rennie,P., Sheppard,P., Matusik,R. and Pinsky,L.  
TITLE Substitution of valine-865 by methionine or leucine in the human androgen receptor causes complete or partial androgen insensitivity, respectively with distinct androgen receptor phenotypes  
JOURNAL Mol. Endocrinol. 7 (1), 37-46 (1993)  
MEDLINE 93188862  
PUBMED 8446106  
REMARK VARIANT CAIS MET-866, AND VARIANT PAIS LEU-866.  
REFERENCE 51 (residues 1 to 919)  
AUTHORS Mowszowicz,I., Lee,H.J., Chen,H.T., Mestayer,C., Portois,M.C., Cabrol,S., Mauvais-Jarvis,P. and Chang,C.  
TITLE A point mutation in the second zinc finger of the DNA-binding domain of the androgen receptor gene causes complete androgen insensitivity in two siblings with receptor-positive androgen resistance  
JOURNAL Mol. Endocrinol. 7 (7), 861-869 (1993)  
MEDLINE 94019395  
PUBMED 8413310  
REMARK VARIANT CAIS HIS-615.  
REFERENCE 52 (residues 1 to 919)  
AUTHORS Culig,Z., Hobisch,A., Cronauer,M.V., Cato,A.C., Hittmair,A., Radmayr,C., Eberle,J., Bartsch,G. and Klocker,H.  
TITLE Mutant androgen receptor detected in an advanced-stage prostatic carcinoma is activated by adrenal androgens and progesterone  
JOURNAL Mol. Endocrinol. 7 (12), 1541-1550 (1993)  
MEDLINE 94195311  
PUBMED 8145761  
REMARK VARIANT PROSTATE CANCER MET-715.  
REFERENCE 53 (residues 1 to 919)  
AUTHORS Lobaccaro,J.-M., Lumbroso,S., Belon,C., Chaussain,J.L., Toublanc,J.E., Leheup,B. and Sultan,C.  
TITLE Androgen receptor (AR) gene mutations in 6 families with androgen insensitivity syndrome (Abstract #114)  
JOURNAL Pediatr. Res. Suppl. 33, S22-S22 (1993)  
REMARK VARIANTS CAIS PHE-581; VAL-743; VAL-754; GLU-767 AND CYS-855.  
REFERENCE 54 (residues 1 to 919)  
AUTHORS Castagnaro,M., Yandell,D.W., Dockhorn-Dworniczak,B., Wolfe,H.J. and Poremba,C.  
TITLE Androgen receptor gene mutations and p53 gene analysis in advanced prostate cancer  
JOURNAL Verh Dtsch Ges Pathol 77, 119-123 (1993)  
MEDLINE 94189145  
PUBMED 7511268  
REMARK VARIANTS PROSTATE CANCER LEU-340 AND GLU-798.  
REFERENCE 55 (residues 1 to 919)  
AUTHORS Schoenberg,M.P., Hakimi,J.M., Wang,S., Bova,G.S., Epstein,J.I., Fischbeck,K.H., Isaacs,W.B., Walsh,P.C. and Barrack,E.R.  
TITLE Microsatellite mutation (CAG24-->18) in the androgen receptor gene in human prostate cancer  
JOURNAL Biochem. Biophys. Res. Commun. 198 (1), 74-80 (1994)  
MEDLINE 94121667  
PUBMED 8292051  
REMARK VARIANT PROSTATE CANCER IN POLY-GLN REGION.

REFERENCE 56 (residues 1 to 919)  
AUTHORS Gaddipati,J.P., McLeod,D.G., Heidenberg,H.B., Sesterhenn,I.A., Finger,M.J., Moul,J.W. and Srivastava,S.  
TITLE Frequent detection of codon 877 mutation in the androgen receptor gene in advanced prostate cancers  
JOURNAL Cancer Res. 54 (11), 2861-2864 (1994)  
MEDLINE 94243798  
PUBMED 8187068  
REMARK VARIANT PROSTATE CANCER ALA-877.  
REFERENCE 57 (residues 1 to 919)  
AUTHORS Lobaccaro,J.M., Belon,C., Lumbroso,S., Olewniczack,G., Carre-Pigeon,F., Job,J.C., Chaussain,J.L., Toublanc,J.E. and Sultan,C.  
TITLE Molecular prenatal diagnosis of partial androgen insensitivity syndrome based on the Hind III polymorphism of the androgen receptor gene  
JOURNAL Clin. Endocrinol. (Oxf) 40 (3), 297-302 (1994)  
MEDLINE 94244054  
PUBMED 7910529  
REMARK VARIANT PAIS TRP-568.  
REFERENCE 58 (residues 1 to 919)  
AUTHORS Lumbroso,S., Lobaccaro,J.M., Belon,C., Amram,S., Bachelard,B., Garandeau,P. and Sultan,C.  
TITLE Molecular prenatal exclusion of familial partial androgen insensitivity (Reifenstein syndrome)  
JOURNAL Eur. J. Endocrinol. 130 (4), 327-332 (1994)  
MEDLINE 94214640  
PUBMED 7909256  
REMARK VARIANT PAIS HIS-840.  
REFERENCE 59 (residues 1 to 919)  
AUTHORS Imasaki,K., Hasegawa,T., Okabe,T., Sakai,Y., Haji,M., Takayanagi,R. and Nawata,H.  
TITLE Single amino acid substitution (840Arg-->His) in the hormone-binding domain of the androgen receptor leads to incomplete androgen insensitivity syndrome associated with a thermolabile androgen receptor  
JOURNAL Eur. J. Endocrinol. 130 (6), 569-574 (1994)  
MEDLINE 94264834  
PUBMED 8205256  
REMARK VARIANT PAIS HIS-840.  
REFERENCE 60 (residues 1 to 919)  
AUTHORS Hiort,O., Klauber,G., Cendron,M., Sinnecker,G.H., Keim,L., Swinger,E., Wolfe,H.J. and Yandell,D.W.  
TITLE Molecular characterization of the androgen receptor gene in boys with hypospadias  
JOURNAL Eur. J. Pediatr. 153 (5), 317-321 (1994)  
MEDLINE 94307287  
PUBMED 8033918  
REMARK VARIANT PAIS VAL-870.  
REFERENCE 61 (residues 1 to 919)  
AUTHORS Schwartz,M., Skovby,F., Mueller,J., Nielsen,O. and Skakkebaek,N.E.  
TITLE Partial androgen insensitivity (PAIS) in a large eskimo kindred caused by a delD690 mutation in the androgen receptor (AR) gene (Abstract #244)  
JOURNAL Horm. Res. 41, 117-117 (1994)  
REMARK VARIANT PAIS ASP-690 DEL.  
REFERENCE 62 (residues 1 to 919)  
AUTHORS Beitel,L.K., Prior,L., Vasiliou,D.M., Gottlieb,B., Kaufman,M., Lumbroso,R., Alvarado,C., McGillivray,B., Trifiro,M. and Pinsky,L.  
TITLE Complete androgen insensitivity due to mutations in the probable

alpha-helical segments of the DNA-binding domain in the human androgen receptor  
JOURNAL Hum. Mol. Genet. 3 (1), 21-27 (1994)  
MEDLINE 94214451  
PUBMED 8162033  
REMARK VARIANTS CAIS PHE-582 DEL; ARG-615 DEL AND HIS-615.  
REFERENCE 63 (residues 1 to 919)  
AUTHORS Hiort,O., Wodtke,A., Struve,D., Zollner,A. and Sinnecker,G.H.  
TITLE Detection of point mutations in the androgen receptor gene using non-isotopic single strand conformation polymorphism analysis.  
German Collaborative Intersex Study Group  
JOURNAL Hum. Mol. Genet. 3 (7), 1163-1166 (1994)  
MEDLINE 95072584  
PUBMED 7981687  
REMARK VARIANTS PAIS SER-582; TYR-604; ALA-708; LEU-754 AND HIS-771, AND VARIANT CAIS TRP-779.  
REFERENCE 64 (residues 1 to 919)  
AUTHORS Baldazzi,L., Baroncini,C., Pirazzoli,P., Balsamo,A., Capelli,M., Marchetti,G., Bernardi,F. and Cacciari,E.  
TITLE Two mutations causing complete androgen insensitivity: a frame-shift in the steroid binding domain and a Cys-->Phe substitution in the second zinc finger of the androgen receptor  
JOURNAL Hum. Mol. Genet. 3 (7), 1169-1170 (1994)  
MEDLINE 95072586  
PUBMED 7981689  
REMARK VARIANT CAIS PHE-601.  
REFERENCE 65 (residues 1 to 919)  
AUTHORS De Bellis,A., Quigley,C.A., Marschke,K.B., el-Awady,M.K., Lane,M.V., Smith,E.P., Sar,M., Wilson,E.M. and French,F.S.  
TITLE Characterization of mutant androgen receptors causing partial androgen insensitivity syndrome  
JOURNAL J. Clin. Endocrinol. Metab. 78 (3), 513-522 (1994)  
MEDLINE 94171952  
PUBMED 8126121  
REMARK VARIANTS PAIS ARG-616; HIS-840 AND MET-889.  
REFERENCE 66 (residues 1 to 919)  
AUTHORS Tsukada,T., Inoue,M., Tachibana,S., Nakai,Y. and Takebe,H.  
TITLE An androgen receptor mutation causing androgen resistance in undervirilized male syndrome  
JOURNAL J. Clin. Endocrinol. Metab. 79 (4), 1202-1207 (1994)  
MEDLINE 95051264  
PUBMED 7962294  
REMARK VARIANT MAIS PHE-790.  
REFERENCE 67 (residues 1 to 919)  
AUTHORS Beitel,L.K., Kazemi-Esfarjani,P., Kaufman,M., Lumbroso,R., DiGeorge,A.M., Killinger,D.W., Trifiro,M.A. and Pinsky,L.  
TITLE Substitution of arginine-839 by cysteine or histidine in the androgen receptor causes different receptor phenotypes in cultured cells and coordinate degrees of clinical androgen resistance  
JOURNAL J. Clin. Invest. 94 (2), 546-554 (1994)  
MEDLINE 94314956  
PUBMED 8040309  
REMARK VARIANTS PAIS CYS-840 AND HIS-840.  
REFERENCE 68 (residues 1 to 919)  
AUTHORS Marcelli,M., Zoppi,S., Wilson,C.M., Griffin,J.E. and McPhaul,M.J.  
TITLE Amino acid substitutions in the hormone-binding domain of the human androgen receptor alter the stability of the hormone receptor complex  
JOURNAL J. Clin. Invest. 94 (4), 1642-1650 (1994)  
MEDLINE 95015035

PUBMED 7929841  
REMARK VARIANTS CAIS, AND VARIANTS PAIS.  
REFERENCE 69 (residues 1 to 919)  
AUTHORS Yong, E.L., Ng, S.C., Roy, A.C., Yun, G. and Ratnam, S.S.  
TITLE Pregnancy after hormonal correction of severe spermatogenic defect due to mutation in androgen receptor gene  
JOURNAL Lancet 344 (8925), 826-827 (1994)  
MEDLINE 94366236  
PUBMED 7993455  
REMARK VARIANT MAIS LYS-727.  
REFERENCE 70 (residues 1 to 919)  
AUTHORS Ris-Stalpers, C., Hoogenboezem, T., Sleddens, H.F.B.M., Verleun-Mooijman, M.C.T., Degenhart, H.J., Drop, S.L.S., Halley, D.J.J., Oosterwijk, J.C., Hodgins, M.B., Trapman, J. and Brinkmann, A.O.  
TITLE A practical approach to the detection of androgen receptor gene mutations and pedigree analysis in families with x-linked androgen insensitivity  
JOURNAL Pediatr. Res. 36 (2), 227-234 (1994)  
MEDLINE 95061051  
PUBMED 7970939  
REMARK VARIANTS CAIS HIS-615 AND LEU-764, AND VARIANTS PAIS VAL-742 AND THR-745.  
REFERENCE 71 (residues 1 to 919)  
AUTHORS Imai, A., Ohno, T., Iida, K., Ohsuye, K., Okano, Y. and Tamaya, T.  
TITLE A frame-shift mutation of the androgen receptor gene in a patient with receptor-negative complete testicular feminization: comparison with a single base substitution in a receptor-reduced incomplete form  
JOURNAL Ann Clin Biochem 32 (Pt 5), 482-486 (1995)  
MEDLINE 96113133  
PUBMED 8830623  
REMARK VARIANT PAIS HIS-840.  
REFERENCE 72 (residues 1 to 919)  
AUTHORS Takahashi, H., Furusato, M., Allsbrook, W.C. Jr., Nishii, H., Wakui, S., Barrett, J.C. and Boyd, J.  
TITLE Prevalence of androgen receptor gene mutations in latent prostatic carcinomas from Japanese men  
JOURNAL Cancer Res. 55 (8), 1621-1624 (1995)  
MEDLINE 95228039  
PUBMED 7712463  
REMARK VARIANTS PROSTATE CANCER.  
REFERENCE 73 (residues 1 to 919)  
AUTHORS Davies, H.R., Hughes, I.A. and Patterson, M.N.  
TITLE Genetic counselling in complete androgen insensitivity syndrome: trinucleotide repeat polymorphisms, single-strand conformation polymorphism and direct detection of two novel mutations in the androgen receptor gene  
JOURNAL Clin. Endocrinol. (Oxf) 43 (1), 69-77 (1995)  
MEDLINE 95368867  
PUBMED 7641413  
REMARK VARIANT CAIS VAL-881.  
REFERENCE 74 (residues 1 to 919)  
AUTHORS Quigley, C.A., De Bellis, A., Marschke, K.B., el-Awady, M.K., Wilson, E.M. and French, F.S.  
TITLE Androgen receptor defects: historical, clinical, and molecular perspectives  
JOURNAL Endocr. Rev. 16 (3), 271-321 (1995)  
MEDLINE 95401910  
PUBMED 7671849

REMARK VARIANTS CAIS SER-705 AND HIS-763, AND VARIANTS PAIS LEU-725; THR-737; HIS-774 AND GLU-798.

REFERENCE 75 (residues 1 to 919)

AUTHORS Quigley,C.A., De Bellis,A., Marschke,K.B., el-Awady,M.K., Wilson,E.M. and French,F.S.

JOURNAL Endocr. Rev. 1995 16, 546-546 (1995)

REMARK ERRATUM.

REFERENCE 76 (residues 1 to 919)

AUTHORS Shkolny,D.L., Brown,T.R., Punnett,H.H., Kaufman,M., Trifiro,M.A. and Pinsky,L.

TITLE Characterization of alternative amino acid substitutions at arginine 830 of the androgen receptor that cause complete androgen insensitivity in three families

JOURNAL Hum. Mol. Genet. 4 (4), 515-521 (1995)

MEDLINE 95359952

PUBMED 7633398

REMARK VARIANTS CAIS LEU-831 AND GLN-831.

REFERENCE 77 (residues 1 to 919)

AUTHORS Belsham,D.D., Pereira,F., Greenberg,C.R., Liao,S. and Wrogemann,K.

TITLE Leu-676-Pro mutation of the androgen receptor causes complete androgen insensitivity syndrome in a large Hutterite kindred

JOURNAL Hum. Mutat. 5 (1), 28-33 (1995)

MEDLINE 95245337

PUBMED 7537149

REMARK VARIANT CAIS PRO-677.

REFERENCE 78 (residues 1 to 919)

AUTHORS Murono,K., Mendonca,B.B., Arnhold,I.J., Rigon,A.C., Migeon,C.J. and Brown,T.R.

TITLE Human androgen insensitivity due to point mutations encoding amino acid substitutions in the androgen receptor steroid-binding domain

JOURNAL Hum. Mutat. 6 (2), 152-162 (1995)

MEDLINE 96047148

PUBMED 7581399

REMARK VARIANT PAIS CYS-763, AND VARIANTS CAIS TRP-779; VAL-807 AND CYS-855.

REFERENCE 79 (residues 1 to 919)

AUTHORS Peterziel,H., Culig,Z., Stober,J., Hobisch,A., Radmayr,C., Bartsch,G., Klocker,H. and Cato,A.C.

TITLE Mutant androgen receptors in prostatic tumors distinguish between amino-acid-sequence requirements for transactivation and ligand binding

JOURNAL Int. J. Cancer 63 (4), 544-550 (1995)

MEDLINE 96074743

PUBMED 7591265

REMARK VARIANT PROSTATE CANCER MET-730.

REFERENCE 80 (residues 1 to 919)

AUTHORS Allera,A., Herbst,M.A., Griffin,J.E., Wilson,J.D., Schweikert,H.U. and McPhaul,M.J.

TITLE Mutations of the androgen receptor coding sequence are infrequent in patients with isolated hypospadias

JOURNAL J. Clin. Endocrinol. Metab. 80 (9), 2697-2699 (1995)

MEDLINE 95403657

PUBMED 7673412

REMARK VARIANT PAIS VAL-568.

REFERENCE 81 (residues 1 to 919)

AUTHORS Elo,J.P., Kvist,L., Leinonen,K., Isomaa,V., Henttu,P., Lukkarinen,O. and Vihko,P.

TITLE Mutated human androgen receptor gene detected in a prostatic cancer patient is also activated by estradiol

JOURNAL J. Clin. Endocrinol. Metab. 80 (12), 3494-3500 (1995)

MEDLINE 96094386  
PUBMED 8530589  
REMARK VARIANT PROSTATE CANCER LEU-726.  
REFERENCE 82 (residues 1 to 919)  
AUTHORS Gast,A., Neuschmid-Kaspar,F., Klocker,H. and Cato,A.C.  
TITLE A single amino acid exchange abolishes dimerization of the androgen receptor and causes Reifenstein syndrome  
JOURNAL Mol. Cell. Endocrinol. 111 (1), 93-98 (1995)  
MEDLINE 95377517  
PUBMED 7649358  
REMARK VARIANT PAIS THR-596.  
REFERENCE 83 (residues 1 to 919)  
AUTHORS Taplin,M.E., Bubley,G.J., Shuster,T.D., Frantz,M.E., Spooner,A.E., Ogata,G.K., Keer,H.N. and Balk,S.P.  
TITLE Mutation of the androgen-receptor gene in metastatic androgen-independent prostate cancer  
JOURNAL N. Engl. J. Med. 332 (21), 1393-1398 (1995)  
MEDLINE 95240676  
PUBMED 7723794  
REMARK VARIANTS PROSTATE CANCER.  
REFERENCE 84 (residues 1 to 919)  
AUTHORS Hiort,O., Sinnecker,G.H., Holterhus,P.M., Nitsche,E.M. and Kruse,K.  
TITLE The clinical and molecular spectrum of androgen insensitivity syndromes  
JOURNAL Am. J. Med. Genet. 63 (1), 218-222 (1996)  
MEDLINE 96298278  
PUBMED 8723113  
REMARK VARIANTS CAIS AND PAIS.  
REFERENCE 85 (residues 1 to 919)  
AUTHORS Tilley,W.D., Buchanan,G., Hickey,T.E. and Bentel,J.M.  
TITLE Mutations in the androgen receptor gene are associated with progression of human prostate cancer to androgen independence  
JOURNAL Clin. Cancer Res. 2 (2), 277-285 (1996)  
MEDLINE 99035018  
PUBMED 9816170  
REMARK VARIANTS PROSTATE CANCER.  
REFERENCE 86 (residues 1 to 919)  
AUTHORS Weidemann,W., Linck,B., Haupt,H., Mentrup,B., Romalo,G., Stockklauser,K., Brinkmann,A.O., Schweikert,H.U. and Spindler,K.D.  
TITLE Clinical and biochemical investigations and molecular analysis of subjects with mutations in the androgen receptor gene  
JOURNAL Clin. Endocrinol. (Oxf) 45 (6), 733-739 (1996)  
MEDLINE 97191382  
PUBMED 9039340  
REMARK VARIANTS PAIS GLN-607; THR-610; LEU-754; HIS-840; THR-842 AND HIS-855, AND VARIANT CAIS MET-866.  
REFERENCE 87 (residues 1 to 919)  
AUTHORS Malmgren,H., Gustavsson,J., Tuvemo,T. and Dahl,N.  
TITLE Rapid detection of a mutation hot-spot in the human androgen receptor  
JOURNAL Clin. Genet. 50 (4), 202-205 (1996)  
MEDLINE 97155085  
PUBMED 9001799  
REMARK VARIANT CAIS CYS-855.  
REFERENCE 88 (residues 1 to 919)  
AUTHORS Bevan,C.L., Brown,B.B., Davies,H.R., Evans,B.A., Hughes,I.A. and Patterson,M.N.  
TITLE Functional analysis of six androgen receptor mutations identified in patients with partial androgen insensitivity syndrome  
JOURNAL Hum. Mol. Genet. 5 (2), 265-273 (1996)

MEDLINE 96422266  
PUBMED 8824883  
REMARK VARIANTS PAIS ILE-742; ILE-780; GLU-798; CYS-840; HIS-855 AND MET-869.  
REFERENCE 89 (residues 1 to 919)  
AUTHORS Choong,C.S., Sturm,M.J., Strophair,J.A., McCulloch,R.K., Tilley,W.D., Leedman,P.J. and Hurley,D.M.  
TITLE Partial androgen insensitivity caused by an androgen receptor mutation at amino acid 907 (Gly-->Arg) that results in decreased ligand binding affinity and reduced androgen receptor messenger ribonucleic acid levels  
JOURNAL J. Clin. Endocrinol. Metab. 81 (1), 236-243 (1996)  
MEDLINE 96141994  
PUBMED 8550758  
REMARK VARIANT PAIS ARG-909.  
REFERENCE 90 (residues 1 to 919)  
AUTHORS Lumbroso,S., Lobaccaro,J.M., Georget,V., Leger,J., Poujol,N., Terouanne,B., Evain-Brion,D., Czernichow,P. and Sultan,C.  
TITLE A novel substitution (Leu707Arg) in exon 4 of the androgen receptor gene causes complete androgen resistance  
JOURNAL J. Clin. Endocrinol. Metab. 81 (5), 1984-1988 (1996)  
MEDLINE 96210890  
PUBMED 8626869  
REMARK VARIANT CAIS ARG-707.  
REFERENCE 91 (residues 1 to 919)  
AUTHORS Rodien,P., Mebarki,F., Mowszowicz,I., Chaussain,J.L., Young,J., Morel,Y. and Schaison,G.  
TITLE Different phenotypes in a family with androgen insensitivity caused by the same M780I point mutation in the androgen receptor gene  
JOURNAL J. Clin. Endocrinol. Metab. 81 (8), 2994-2998 (1996)  
MEDLINE 96320094  
PUBMED 8768864  
REMARK VARIANT PAIS/CAIS ILE-780.  
REFERENCE 92 (residues 1 to 919)  
AUTHORS Choong,C.S., Quigley,C.A., French,F.S. and Wilson,E.M.  
TITLE A novel missense mutation in the amino-terminal domain of the human androgen receptor gene in a family with partial androgen insensitivity syndrome causes reduced efficiency of protein translation  
JOURNAL J. Clin. Invest. 98 (6), 1423-1431 (1996)  
MEDLINE 96420610  
PUBMED 8823308  
REMARK VARIANT PAIS LYS-2.  
REFERENCE 93 (residues 1 to 919)  
AUTHORS Bruggenwirth,H.T., Boehmer,A.L., Verleun-Mooijman,M.C., Hoogenboezem,T., Kleijer,W.J., Otten,B.J., Trapman,J. and Brinkmann,A.O.  
TITLE Molecular basis of androgen insensitivity  
JOURNAL J. Steroid Biochem. Mol. Biol. 58 (5-6), 569-575 (1996)  
MEDLINE 97078859  
PUBMED 8918984  
REMARK VARIANT CAIS ASP-573.  
REFERENCE 94 (residues 1 to 919)  
AUTHORS Sutherland,R.W., Wiener,J.S., Hicks,J.P., Marcelli,M., Gonzales,E.T. Jr., Roth,D.R. and Lamb,D.J.  
TITLE Androgen receptor gene mutations are rarely associated with isolated penile hypospadias  
JOURNAL J. Urol. 156 (2 Pt 2), 828-831 (1996)  
MEDLINE 96289452  
PUBMED 8683794

REMARK VARIANT MAIS SER-548.  
REFERENCE 95 (residues 1 to 919)  
AUTHORS Lobaccaro,J.M., Poujol,N., Chiche,L., Lumbroso,S., Brown,T.R. and Sultan,C.  
TITLE Molecular modeling and in vitro investigations of the human androgen receptor DNA-binding domain: application for the study of two mutations  
JOURNAL Mol. Cell. Endocrinol. 116 (2), 137-147 (1996)  
MEDLINE 96231386  
PUBMED 8647313  
REMARK VARIANT CAIS PRO-616.  
REFERENCE 96 (residues 1 to 919)  
AUTHORS Imasaki,K., Okabe,T., Murakami,H., Tanaka,Y., Haji,M., Takayanagi,R. and Nawata,H.  
TITLE Androgen insensitivity syndrome due to new mutations in the DNA-binding domain of the androgen receptor  
JOURNAL Mol. Cell. Endocrinol. 120 (1), 15-24 (1996)  
MEDLINE 96405605  
PUBMED 8809734  
REMARK VARIANT CAIS PHE-579, AND VARIANT PAIS TYR-582.  
REFERENCE 97 (residues 1 to 919)  
AUTHORS Evans,B.A., Harper,M.E., Daniells,C.E., Watts,C.E., Matenhelia,S., Green,J. and Griffiths,K.  
TITLE Low incidence of androgen receptor gene mutations in human prostatic tumors using single strand conformation polymorphism analysis  
JOURNAL Prostate 28 (3), 162-171 (1996)  
MEDLINE 96235947  
PUBMED 8628719  
REMARK VARIANT PROSTATE CANCER GLU-798.  
REFERENCE 98 (residues 1 to 919)  
AUTHORS Suzuki,H., Akakura,K., Komiya,A., Aida,S., Akimoto,S. and Shimazaki,J.  
TITLE Codon 877 mutation in the androgen receptor gene in advanced prostate cancer: relation to antiandrogen withdrawal syndrome  
JOURNAL Prostate 29 (3), 153-158 (1996)  
MEDLINE 96424622  
PUBMED 8827083  
REMARK VARIANT PROSTATE CANCER ALA-877.  
REFERENCE 99 (residues 1 to 919)  
AUTHORS Boehmer,A.L., Brinkmann,A.O., Niermeijer,M.F., Bakker,L., Halley,D.J. and Drop,S.L.  
TITLE Germ-line and somatic mosaicism in the androgen insensitivity syndrome: implications for genetic counseling  
JOURNAL Am. J. Hum. Genet. 60 (4), 1003-1006 (1997)  
MEDLINE 97260430  
PUBMED 9106550  
REMARK VARIANT PAIS HIS-855.  
REFERENCE 100 (residues 1 to 919)  
AUTHORS Koivisto,P., Kononen,J., Palmberg,C., Tammela,T., Hyytinen,E., Isola,J., Trapman,J., Cleutjens,K., Noordzij,A., Visakorpi,T. and Kallioniemi,O.-P.  
TITLE Androgen receptor gene amplification: a possible molecular mechanism for androgen deprivation therapy failure in prostate cancer  
JOURNAL Cancer Res. 57 (2), 314-319 (1997)  
MEDLINE 97153285  
PUBMED 9000575  
REMARK VARIANT PROSTATE CANCER ALA-683.  
REFERENCE 101 (residues 1 to 919)

AUTHORS Tincello,D.G., Saunders,P.T., Hodgins,M.B., Simpson,N.B., Edwards,C.R., Hargreaves,T.B. and Wu,F.C.  
TITLE Correlation of clinical, endocrine and molecular abnormalities with in vivo responses to high-dose testosterone in patients with partial androgen insensitivity syndrome  
JOURNAL Clin. Endocrinol. (Oxf) 46 (4), 497-506 (1997)  
MEDLINE 97340104  
PUBMED 9196614  
REMARK VARIANTS PAIS LYS-608 AND GLY-772.  
REFERENCE 102 (residues 1 to 919)  
AUTHORS Essawi,M., Gad,Y.Z., el-Rouby,O., Temtamy,S.A., Sabour,Y.A. and el-Awady,M.K.  
TITLE Molecular analysis of androgen resistance syndromes in Egyptian patients  
JOURNAL Dis. Markers 13 (2), 99-105 (1997)  
MEDLINE 97303883  
PUBMED 9160185  
REMARK VARIANT PAIS MET-889.  
REFERENCE 103 (residues 1 to 919)  
AUTHORS Sinnecker,G.H., Hiort,O., Nitsche,E.M., Holterhus,P.M. and Kruse,K.  
TITLE Functional assessment and clinical classification of androgen sensitivity in patients with mutations of the androgen receptor gene. German Collaborative Intersex Study Group  
JOURNAL Eur. J. Pediatr. 156 (1), 7-14 (1997)  
MEDLINE 97159966  
PUBMED 9007482  
REMARK VARIANT CAIS TRP-779.  
REFERENCE 104 (residues 1 to 919)  
AUTHORS Jakubiczka,S., Nedel,S., Werder,E.A., Schleiermacher,E., Theile,U., Wolff,G. and Wieacker,P.  
TITLE Mutations of the androgen receptor gene in patients with complete androgen insensitivity  
JOURNAL Hum. Mutat. 9 (1), 57-61 (1997)  
MEDLINE 97144184  
PUBMED 8990010  
REMARK VARIANTS CAIS VAL-749; CYS-774; ILE-780 AND SER-794.  
REFERENCE 105 (residues 1 to 919)  
AUTHORS Watanabe,M., Ushijima,T., Shiraishi,T., Yatani,R., Shimazaki,J., Kotake,T., Sugimura,T. and Nagao,M.  
TITLE Genetic alterations of androgen receptor gene in Japanese human prostate cancer  
JOURNAL Jpn J Clin Oncol 27 (6), 389-393 (1997)  
MEDLINE 98100812  
PUBMED 9438000  
REMARK VARIANTS PROSTATE CANCER IN POLY-GLN REGION; HIS-701 AND ARG-910.  
REFERENCE 106 (residues 1 to 919)  
AUTHORS Wang,C. and Uchida,T.  
TITLE Androgen receptor gene mutations in prostate cancer  
JOURNAL Jpn. J. Urol. 88, 550-556 (1997)  
REMARK VARIANT PROSTATE CANCER GLN-629.  
REFERENCE 107 (residues 1 to 919)  
AUTHORS Komori,S., Sakata,K., Tanaka,H., Shima,H. and Koyama,K.  
TITLE DNA analysis of the androgen receptor gene in two cases with complete androgen insensitivity syndrome  
JOURNAL J Obstet Gynaecol Res 23 (3), 277-281 (1997)  
MEDLINE 97398857  
PUBMED 9255042  
REMARK VARIANTS CAIS ARG-194 AND CYS-855.  
REFERENCE 108 (residues 1 to 919)  
AUTHORS Albers,N., Ulrichs,C., Gluer,S., Hiort,O., Sinnecker,G.H.,

Mildenberger,H. and Brodehl,J.  
TITLE Etiologic classification of severe hypospadias: implications for prognosis and management  
JOURNAL J. Pediatr. 131 (3), 386-392 (1997)  
MEDLINE 97469997  
PUBMED 9329414  
REMARK VARIANTS PAIS ALA-708 AND GLY-870.  
REFERENCE 109 (residues 1 to 919)  
AUTHORS Ko,T.M., Yang,Y.S., Wu,M.Y., Kao,C.H., Hsu,P.M., Chuang,S.M. and Lee,T.Y.  
TITLE Complete androgen insensitivity syndrome. Molecular characterization in two Chinese women  
JOURNAL J Reprod Med 42 (7), 424-428 (1997)  
MEDLINE 97396769  
PUBMED 9252933  
REMARK VARIANTS CAIS ASN-732 AND THR-765.  
REFERENCE 110 (residues 1 to 919)  
AUTHORS Bevan,C.L., Hughes,I.A. and Patterson,M.N.  
TITLE Wide variation in androgen receptor dysfunction in complete androgen insensitivity syndrome  
JOURNAL J. Steroid Biochem. Mol. Biol. 61 (1-2), 19-26 (1997)  
MEDLINE 97466866  
PUBMED 9328206  
REMARK VARIANTS CAIS ASP-750; PHE-762; THR-765; ASN-864 AND PHE-907.  
REFERENCE 111 (residues 1 to 919)  
AUTHORS Radmayr,C., Culig,Z., Glatzl,J., Neuschmid-Kaspar,F., Bartsch,G. and Klocker,H.  
TITLE Androgen receptor point mutations as the underlying molecular defect in 2 patients with androgen insensitivity syndrome  
JOURNAL J. Urol. 158 (4), 1553-1556 (1997)  
MEDLINE 97445885  
PUBMED 9302173  
REMARK VARIANT PAIS GLY-703, AND VARIANT CAIS LEU-916.  
REFERENCE 112 (residues 1 to 919)  
AUTHORS Komori,S., Kasumi,H., Sakata,K., Tanaka,H., Hamada,K. and Koyama,K.  
TITLE Molecular analysis of the androgen receptor gene in 4 patients with complete androgen insensitivity  
JOURNAL Arch. Gynecol. Obstet. 261 (2), 95-100 (1998)  
MEDLINE 98206019  
PUBMED 9544375  
REMARK VARIANTS CAIS CYS-571; GLN-752 AND CYS-774.  
REFERENCE 113 (residues 1 to 919)  
AUTHORS Cabral,D.F., Maciel-Guerra,A.T. and Hackel,C.  
TITLE Mutations of androgen receptor gene in Brazilian patients with male pseudohermaphroditism  
JOURNAL Braz. J. Med. Biol. Res. 31 (6), 775-778 (1998)  
MEDLINE 98363978  
PUBMED 9698822  
REMARK VARIANTS CAIS HIS-615 AND GLN-752.  
REFERENCE 114 (residues 1 to 919)  
AUTHORS Wang,Q., Ghadessy,F.J. and Yong,E.L.  
TITLE Analysis of the transactivation domain of the androgen receptor in patients with male infertility  
JOURNAL Clin. Genet. 54 (3), 185-192 (1998)  
MEDLINE 99002768  
PUBMED 9788719  
REMARK VARIANT MAIS ARG-214.  
REFERENCE 115 (residues 1 to 919)  
AUTHORS Tanaka,H., Komori,S., Sakata,K., Shima,H. and Koyama,K.  
TITLE One additional mutation at exon A amplifies thermolability of

androgen receptor in a case with complete androgen insensitivity syndrome  
JOURNAL Gynecol Endocrinol 12 (2), 75-82 (1998)  
MEDLINE 98273339  
PUBMED 9610419  
REMARK VARIANTS CAIS PRO-255 AND ALA-820.  
REFERENCE 116 (residues 1 to 919)  
AUTHORS Lundberg Giwercman,Y., Nikoshkov,A., Lindsten,K., Bystroem,B., Pousette,A., Chibalin,A.V., Arvidsson,S., Tiulpakov,A., Semitcheva,T.V., Peterkova,V., Hagenfeldt,K., Ritzen,E.M. and Wedell,A.  
TITLE Functional characterisation of mutations in the ligand-binding domain of the androgen receptor gene in patients with androgen insensitivity syndrome  
JOURNAL Hum. Genet. 103 (4), 529-531 (1998)  
MEDLINE 99072324  
PUBMED 9856504  
REMARK VARIANTS CAIS THR-765; TYR-784 AND THR-895, AND VARIANT PAIS GLY-840.  
REFERENCE 117 (residues 1 to 919)  
AUTHORS Doerk,T., Schnieders,F., Jakubiczka,S., Wieacker,P., Schroeder-Kurth,T. and Schmidtke,J.  
TITLE A new missense substitution at a mutational hot spot of the androgen receptor in siblings with complete androgen insensitivity syndrome  
JOURNAL Hum. Mutat. 11, 337-339 (1998)  
REMARK VARIANT CAIS VAL-695.  
REFERENCE 118 (residues 1 to 919)  
AUTHORS Nordenskjoejd,A. and Soederhaell,S.  
TITLE An androgen receptor gene mutation (A645D) in a boy with a normal phenotype  
JOURNAL Hum. Mutat. 11, 339-339 (1998)  
REMARK VARIANT ASP-645.  
REFERENCE 119 (residues 1 to 919)  
AUTHORS Knoke,I., Jakubiczka,S., Rohrer,T., Hanemann,B., Werder,E.A. and Wieacker,P.  
TITLE Single amino acid substitution in the hormone-binding domain of the androgen receptor in a family with complete androgen insensitivity syndrome (CAIS)  
JOURNAL Hum. Mutat. 12, 220-220 (1998)  
REMARK VARIANT CAIS LEU-892.  
REFERENCE 120 (residues 1 to 919)  
AUTHORS Weidemann,W., Peters,B., Romalo,G., Spindler,K.D. and Schweikert,H.U.  
TITLE Response to androgen treatment in a patient with partial androgen insensitivity and a mutation in the deoxyribonucleic acid-binding domain of the androgen receptor  
JOURNAL J. Clin. Endocrinol. Metab. 83 (4), 1173-1176 (1998)  
MEDLINE 98202136  
PUBMED 9543136  
REMARK VARIANT PAIS GLN-607.  
REFERENCE 121 (residues 1 to 919)  
AUTHORS Georget,V., Terouanne,B., Lumbroso,S., Nicolas,J.C. and Sultan,C.  
TITLE Trafficking of androgen receptor mutants fused to green fluorescent protein: a new investigation of partial androgen insensitivity syndrome  
JOURNAL J. Clin. Endocrinol. Metab. 83 (10), 3597-3603 (1998)  
MEDLINE 98439607  
PUBMED 9768671  
REMARK VARIANTS PAIS VAL-743 AND CYS-840.

REFERENCE 122 (residues 1 to 919)  
AUTHORS Wang,Q., Ghadessy,F.J., Trounson,A., de Kretser,D., McLachlan,R., Ng,S.C. and Yong,E.L.  
TITLE Azoospermia associated with a mutation in the ligand-binding domain of an androgen receptor displaying normal ligand binding, but defective trans-activation  
JOURNAL J. Clin. Endocrinol. Metab. 83 (12), 4303-4309 (1998)  
MEDLINE 99067093  
PUBMED 9851768  
REMARK VARIANT MAIS GLU-798.  
REFERENCE 123 (residues 1 to 919)  
AUTHORS Hiort,O., Sinnecker,G.H., Holterhus,P.M., Nitsche,E.M. and Kruse,K.  
TITLE Inherited and de novo androgen receptor gene mutations: investigation of single-case families  
JOURNAL J. Pediatr. 132 (6), 939-943 (1998)  
MEDLINE 98291052  
PUBMED 9627582  
REMARK VARIANTS CAIS AND PAIS.  
REFERENCE 124 (residues 1 to 919)  
AUTHORS Yong,E.L., Tut,T.G., Ghadessy,F.J., Prins,G. and Ratnam,S.S.  
TITLE Partial androgen insensitivity and correlations with the predicted three dimensional structure of the androgen receptor ligand-binding domain  
JOURNAL Mol. Cell. Endocrinol. 137 (1), 41-50 (1998)  
MEDLINE 98268743  
PUBMED 9607727  
REMARK VARIANT PAIS THR-758.  
REFERENCE 125 (residues 1 to 919)  
AUTHORS Knoke,I., Jakubiczka,S., Lehnert,H. and Wieacker,P.  
TITLE A new point mutation of the androgen receptor gene in a patient with partial androgen resistance and severe oligozoospermia  
JOURNAL Andrologia 31 (4), 199-201 (1999)  
MEDLINE 99399430  
PUBMED 10470409  
REMARK VARIANT PAIS LEU-911.  
REFERENCE 126 (residues 1 to 919)  
AUTHORS Taplin,M.E., Bubley,G.J., Ko,Y.J., Small,E.J., Upton,M., Rajeshkumar,B. and Balk,S.P.  
TITLE Selection for androgen receptor mutations in prostate cancers treated with androgen antagonist  
JOURNAL Cancer Res. 59 (11), 2511-2515 (1999)  
MEDLINE 99290631  
PUBMED 10363963  
REMARK VARIANTS PROSTATE CANCER ALA-877 AND ASN-890.  
REFERENCE 127 (residues 1 to 919)  
AUTHORS Melo,K.F.S., Latronico,A.C., Costa,E.M.F., Billerbeck,A.E.C., Mendonca,B.B. and Arnhold,I.J.P.  
TITLE A novel point mutation (R840S) in the androgen receptor in a Brazilian family with partial androgen insensitivity syndrome  
JOURNAL Hum. Mutat. 14, 353-353 (1999)  
REMARK VARIANT PAIS SER-840.  
REFERENCE 128 (residues 1 to 919)  
AUTHORS Gottlieb,B., Vasiliou,D.M., Lumbroso,R., Beitel,L.K., Pinsky,L. and Trifiro,M.A.  
TITLE Analysis of exon 1 mutations in the androgen receptor gene  
JOURNAL Hum. Mutat. 14 (6), 527-539 (1999)  
MEDLINE 20040031  
PUBMED 10571951  
REMARK VARIANTS CAIS ARG-390 AND ARG-443.  
REFERENCE 129 (residues 1 to 919)

AUTHORS Chen,C.P., Chern,S.R., Wang,T.Y., Wang,W., Wang,K.L. and Jeng,C.J.  
TITLE Androgen receptor gene mutations in 46,XY females with germ cell tumours  
JOURNAL Hum. Reprod. 14 (3), 664-670 (1999)  
MEDLINE 99236881  
PUBMED 10221692  
REMARK VARIANT PAIS GLN-607, AND VARIANT CAIS LYS-681.  
REFERENCE  
AUTHORS Kanayama,H., Naroda,T., Inoue,Y., Kurokawa,Y. and Kagawa,S.  
TITLE A case of complete testicular feminization: laparoscopic orchietomy and analysis of androgen receptor gene mutation  
JOURNAL Int J Urol 6 (6), 327-330 (1999)  
MEDLINE 99332382  
PUBMED 10404311  
REMARK VARIANT CAIS LEU-892.  
REFERENCE  
AUTHORS Shkolny,D.L., Beitel,L.K., Ginsberg,J., Pekelis,G., Arbour,L., Pinsky,L. and Trifiro,M.A.  
TITLE Discordant measures of androgen-binding kinetics in two mutant androgen receptors causing mild or partial androgen insensitivity, respectively  
JOURNAL J. Clin. Endocrinol. Metab. 84 (2), 805-810 (1999)  
MEDLINE 99145056  
PUBMED 10022458  
REMARK VARIANT PAIS ALA-772, AND VARIANT MAIS GLY-871.  
REFERENCE  
AUTHORS Wallen,M.J., Linja,M., Kaartinen,K., Schleutker,J. and Visakorpi,T.  
TITLE Androgen receptor gene mutations in hormone-refractory prostate cancer  
JOURNAL J. Pathol. 189 (4), 559-563 (1999)  
MEDLINE 20096941  
PUBMED 10629558  
REMARK VARIANTS PROSTATE CANCER IN POLY-GLN REGION AND ALA-683.  
REFERENCE  
AUTHORS Zhao,X.Y., Boyle,B., Krishnan,A.V., Navone,N.M., Peehl,D.M. and Feldman,D.  
TITLE Two mutations identified in the androgen receptor of the new human prostate cancer cell line MDA PCa 2a  
JOURNAL J. Urol. 162 (6), 2192-2199 (1999)  
MEDLINE 20034906  
PUBMED 10569618  
REMARK VARIANTS PROSTATE CANCER HIS-701 AND ALA-877.  
REFERENCE  
AUTHORS Ong,Y.C., Wong,H.B., Adaikan,G. and Yong,E.L.  
TITLE Directed pharmacological therapy of ambiguous genitalia due to an androgen receptor gene mutation  
JOURNAL Lancet 354 (9188), 1444-1445 (1999)  
MEDLINE 20009021  
PUBMED 10543676  
REMARK VARIANT PAIS THR-807.  
REFERENCE  
AUTHORS Peters,I., Weidemann,W., Romalo,G., Knorr,D., Schweikert,H.U. and Spindler,K.D.  
TITLE An androgen receptor mutation in the direct vicinity of the proposed C-terminal alpha-helix of the ligand binding domain containing the AF-2 transcriptional activating function core is associated with complete androgen insensitivity  
JOURNAL Mol. Cell. Endocrinol. 148 (1-2), 47-53 (1999)  
MEDLINE 99236959  
PUBMED 10221770

REMARK VARIANT CAIS LEU-892.  
REFERENCE 136 (residues 1 to 919)  
AUTHORS Nazareth,L.V., Stenoien,D.L., Bingman,W.E. III, James,A.J., Wu,C., Zhang,Y., Edwards,D.P., Mancini,M., Marcelli,M., Lamb,D.J. and Weigel,N.L.  
TITLE A C619Y mutation in the human androgen receptor causes inactivation and mislocalization of the receptor with concomitant sequestration of SRC-1 (steroid receptor coactivator 1)  
JOURNAL Mol. Endocrinol. 13 (12), 2065-2075 (1999)  
MEDLINE 20065641  
PUBMED 10598582  
REMARK VARIANT PROSTATE CANCER TYR-619.  
REFERENCE 137 (residues 1 to 919)  
AUTHORS Nazareth,L.V., Stenoien,D.L., Bingman,W.E. III, James,A.J., Wu,C., Zhang,Y., Edwards,D.P., Mancini,M., Marcelli,M., Lamb,D.J. and Weigel,N.L.  
JOURNAL Mol. Endocrinol. 14, 544-544 (2000)  
REMARK ERRATUM.  
REFERENCE 138 (residues 1 to 919)  
AUTHORS Holterhus,P.M., Wiebel,J., Sinnecker,G.H., Bruggenwirth,H.T., Sippell,W.G., Brinkmann,A.O., Kruse,K. and Hiort,O.  
TITLE Clinical and molecular spectrum of somatic mosaicism in androgen insensitivity syndrome  
JOURNAL Pediatr. Res. 46 (6), 684-690 (1999)  
MEDLINE 20055969  
PUBMED 10590024  
REMARK VARIANT PAIS THR-596.  
REFERENCE 139 (residues 1 to 919)  
AUTHORS Yaegashi,N., Uehara,S., Senoo,M., Sato,J., Fujiwara,J., Funato,T., Sasaki,T. and Yajima,A.  
TITLE Point mutations in the steroid-binding domain of the androgen receptor gene of five Japanese patients with androgen insensitivity syndrome  
JOURNAL Tohoku J. Exp. Med. 187 (3), 263-272 (1999)  
MEDLINE 99385665  
PUBMED 10458483  
REMARK VARIANT PAIS PHE-812, AND VARIANT CAIS GLN-831.  
REFERENCE 140 (residues 1 to 919)  
AUTHORS Nordenskjold,A., Friedman,E., Tapper-Persson,M., Soderhall,C., Leviav,A., Svensson,J. and Anvret,M.  
TITLE Screening for mutations in candidate genes for hypospadias  
JOURNAL Urol. Res. 27 (1), 49-55 (1999)  
MEDLINE 99190574  
PUBMED 10092153  
REMARK VARIANTS PAIS THR-597 AND LEU-725.  
REFERENCE 141 (residues 1 to 919)  
AUTHORS Marcelli,M., Ittmann,M., Mariani,S., Sutherland,R.W., Nigam,R., Murthy,L., Zhao,Y., DiConcini,D., Puxeddu,E., Esen,A., Eastham,J., Weigel,N.L. and Lamb,D.J.  
TITLE Androgen receptor mutations in prostate cancer  
JOURNAL Cancer Res. 60 (4), 944-949 (2000)  
MEDLINE 20168626  
PUBMED 10706109  
REMARK VARIANTS PROSTATE CANCER ALA-575; ARG-580; VAL-586; TYR-619; ALA-757 AND GLY-846.  
REFERENCE 142 (residues 1 to 919)  
AUTHORS Ahmed,S.F., Cheng,A., Dovey,L., Hawkins,J.R., Martin,H., Rowland,J., Shimura,N., Tait,A.D. and Hughes,I.A.  
TITLE Phenotypic features, androgen receptor binding, and mutational analysis in 278 clinical cases reported as androgen insensitivity

syndrome  
JOURNAL J. Clin. Endocrinol. Metab. 85 (2), 658-665 (2000)  
MEDLINE 20152731  
PUBMED 10690872  
REMARK VARIANTS CAIS AND PAIS.  
REFERENCE 143 (residues 1 to 919)  
AUTHORS Chavez,B., Mendez,J.P., Ulloa-Aguirre,A., Larrea,F. and Vilchis,F.  
TITLE Eight novel mutations of the androgen receptor gene in patients with androgen insensitivity syndrome  
JOURNAL J. Hum. Genet. 46 (10), 560-565 (2001)  
MEDLINE 21470699  
PUBMED 11587068  
REMARK VARIANTS PAIS THR-682 AND GLU-711, VARIANTS CAIS GLU-743; VAL-827 AND ARG-874, AND VARIANT MAIS TYR-879.  
REFERENCE 144 (residues 1 to 919)  
AUTHORS Sills,E.S., Sholes,T.E., Perloe,M., Kaplan,C.R., Davis,J.G. and Tucker,M.J.  
TITLE Characterization of a novel receptor mutation A-->T at exon 4 in complete androgen insensitivity syndrome and a carrier sibling via bidirectional polymorphism sequence analysis  
JOURNAL Int. J. Mol. Med. 9 (1), 45-48 (2002)  
MEDLINE 21610990  
PUBMED 11744994  
REMARK VARIANT CAIS TYR-705.

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COMMENT  
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[FUNCTION] THE STEROID HORMONES AND THEIR RECEPTORS ARE INVOLVED IN THE REGULATION OF EUKARYOTIC GENE EXPRESSION AND AFFECT CELLULAR PROLIFERATION AND DIFFERENTIATION IN TARGET TISSUES.  
[SUBUNIT] Binds DNA as a homodimer. The AR N-terminal poly-Gln region binds RAN resulting in enhancement of AR-mediated transactivation. RAN binding decreases as the poly-Gln length increases.  
[SUBCELLULAR LOCATION] Nuclear.  
[DOMAIN] Composed of three domains: a modulating N-terminal domain, a DNA-binding domain and a C-terminal steroid-binding domain.  
[POLYMORPHISM] The poly-Gln region of AR is highly polymorphic and the number of Gln varies in the population (from 17 to 26). A smaller size of the poly-Gln region may be associated with the development of prostate cancer.  
[POLYMORPHISM] The poly-Gly region of AR is also polymorphic and ranges from 24 to 31 Gly.  
[DISEASE] DEFECTS IN AR ARE THE CAUSE OF ANDROGEN INSENSIBILITY SYNDROME (AIS), PREVIOUSLY KNOWN AS TESTICULAR FEMINIZATION SYNDROME (TFM). IT CAN BE COMPLETE (CAIS) WHEN EXTERNAL GENITALIA ARE PHENOTYPICALLY FEMALE; OR PARTIAL (PAIS) WHEN EXTERNAL GENITALIA ARE SUBSTANTIALLY AMBIGUOUS OR MILD (MAIS) WHEN EXTERNAL GENITALIA ARE NORMAL MALE OR NEARLY SO.  
[DISEASE] DEFECTS IN AR ARE THE CAUSE OF X-LINKED SPINAL AND BULBAR MUSCULAR ATROPHY (SBMA) (ALSO KNOWN AS KENNEDY'S DISEASE). IN SBMA PATIENTS THE NUMBER OF GLN RANGES FROM 40 TO 52. LONGER EXPANSIONS RESULT IN EARLIER ONSET AND MORE SEVERE CLINICAL MANIFESTATIONS OF THE DISEASE.  
[DISEASE] DEFECTS IN AR MAY PLAY A ROLE IN METASTATIC PROSTATE CANCER. THE MUTATED RECEPTOR STIMULATES PROSTATE GROWTH AND

METASTASES DEVELOPMENT DESPITE OF ANDROGEN ABLATION. THIS TREATMENT CAN REDUCE PRIMARY AND METASTATIC LESIONS PROBABLY BY INDUCING APOPTOSIS OF TUMOR CELLS WHEN THEY EXPRESS THE WILD-TYPE RECEPTOR. [DISEASE] DEFECTS IN AR MAY BE THE CAUSE OF INFERTILITY MALE SYNDROME. IT IS CHARACTERIZED BY AZOOSPERMIA, ELEVATED TESTOSTERONE AND LUTEINIZING HORMONE PLASMA LEVELS AND AN ABNORMAL ANDROGEN RECEPTOR.

[MISCELLANEOUS] In the absence of ligand, steroid hormone receptors are thought to be weakly associated with nuclear components; hormone binding greatly increases receptor affinity. The hormone-receptor complex appears to recognize discrete DNA sequences upstream of transcriptional start sites.

[SIMILARITY] Belongs to the nuclear hormone receptor family. NR3 subfamily.

[DATABASE] NAME=Androgen receptor gene mutations database;  
WWW='<http://www.mcgill.ca/androgendb/>';  
FTP='ftp://ftp.ebi.ac.uk/pub/databases/androgen'.

**FEATURES**

	Location/Qualifiers
<u>source</u>	1..919 /organism="Homo sapiens" /mol_type="unassigned DNA" /db_xref="taxon:9606"
<u>gene</u>	1..919 /gene="AR" /note="synonyms: NR3C4, DHTR"
<u>Protein</u>	1..919 /gene="AR" /product="Androgen receptor"
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<u>Region</u>	2 /gene="AR" /region_name="Variant" /note="E -> K (IN PAIS). /FTId=VAR_004679."
<u>Region</u>	54..57 /gene="AR" /region_name="Domain" /note="POLY-LEU."
<u>Region</u>	54 /gene="AR" /region_name="Variant" /note="L -> S (IN PROSTATE CANCER). /FTId=VAR_004680."
<u>Region</u>	57 /gene="AR" /region_name="Variant" /note="L -> Q (IN PROSTATE CANCER). /FTId=VAR_004681."
<u>Region</u>	58..89 /gene="AR" /region_name="Domain" /note="GLN-RICH."
<u>Region</u>	58..78 /gene="AR" /region_name="Domain" /note="POLY-GLN."
<u>Region</u>	64 /gene="AR" /region_name="Variant" /note="Q -> R (IN PROSTATE CANCER). /FTId=VAR_009711."

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/region\_name="Domain"  
/note="POLY-GLN."  
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/region\_name="Variant"  
/note="K -> R (IN PROSTATE CANCER). /FTId=VAR\_009713."  
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/region\_name="Domain"  
/note="POLY-GLN."  
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/region\_name="Variant"  
/note="Q -> R (IN CAIS). /FTId=VAR\_009224."  
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/region\_name="Variant"  
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/region\_name="Conflict"  
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/region\_name="Variant"  
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CAPACITY). /FTId=VAR\_009715."  
Region 255  
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/region\_name="Variant"  
/note="L -> P (IN CAIS). /FTId=VAR\_009225."  
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/region\_name="Variant"  
/note="P -> S (IN PROSTATE CANCER). /FTId=VAR\_009717."  
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/note="P -> L (IN PROSTATE CANCER). /FTId=VAR\_009718."  
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Region /note="POLY-PRO."  
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Region 390 /gene="AR"  
/region\_name="Variant"  
/note="P -> S (IN MAIS). /FTId=VAR\_009227."  
Region 396..402 /gene="AR"  
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/note="POLY-ALA."  
Region 443 /gene="AR"  
/region\_name="Variant"  
/note="Q -> R (IN CAIS; MIGHT BE A POLYMORPHISM).  
/FTId=VAR\_009228."  
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/region\_name="Domain"  
/note="POLY-GLY."  
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/region\_name="Variant"  
/note="MISSING. /FTId=VAR\_004683."  
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/region\_name="Conflict"  
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/region\_name="Variant"  
/note="L -> F (IN PAIS). /FTId=VAR\_009721."  
Region 548 /gene="AR"  
/region\_name="Variant"  
/note="P -> S (IN MAIS). /FTId=VAR\_009722."  
Site 559..624 /gene="AR"  
/site\_type="DNA binding"  
/note="NUCLEAR RECEPTOR-TYPE."  
Region 559..579 /gene="AR"  
/region\_name="Zinc finger region"  
/note="C4-TYPE."  
Region 559 /gene="AR"  
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Region

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/region\_name="Variant"  
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568  
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/region\_name="Variant"  
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571  
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/region\_name="Variant"  
/note="Y -> C (IN CAIS). /FTId=VAR\_009727."  
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573  
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/region\_name="Variant"  
/note="A -> D (IN CAIS; DEFECTIVE DNA BINDING AND  
TRANSACTIVATION). /FTId=VAR\_009728."  
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/region\_name="Variant"  
/note="L -> P (IN PROSTATE CANCER). /FTId=VAR\_009729."  
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/region\_name="Variant"  
/note="T -> A (IN PROSTATE CANCER). /FTId=VAR\_009730."  
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576  
/gene="AR"  
/region\_name="Variant"  
/note="C -> F (IN CAIS; LACK OF DNA BINDING).  
/FTId=VAR\_009731."  
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576  
/gene="AR"  
/region\_name="Variant"  
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Region  
579  
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581  
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Region  
582  
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Region  
582  
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Region

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582  
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585  
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586  
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/FTId=VAR\_009741."  
587  
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595..619  
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596  
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/FTId=VAR\_009743."  
597  
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WITH P-617 IN A PAIS PATIENT; PARTIALLY RESTORES  
DNA-BINDING ACTIVITY OF P-617 MUTANT RECEPTORS).  
/FTId=VAR\_009744."  
597  
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601  
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604  
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607  
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/FTId=VAR\_004684."  
608  
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NUCLEAR LOCALIZATION). /FTId=VAR\_004685."  
610  
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Region

611  
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Region  
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Region  
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615  
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Region  
616  
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/note="R -> P (IN CAIS). /FTId=VAR\_009752."  
Region  
616  
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/note="L -> P (IN CAIS). /FTId=VAR\_009753."  
Region  
617  
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Region  
619  
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629  
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Region  
630  
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Region  
634  
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Region  
645  
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Region  
647  
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Region  
664  
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Region 672  
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Region 675  
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Region 681  
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Region 683  
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Region 684  
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Region 686  
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Region 690..919  
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Region 692  
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Region 695  
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Region 700  
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Region 701  
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Region 701  
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Region 703  
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Region 705  
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Region 707  
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Region 708  
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Region 710  
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Region                    /note="R -> T (IN CAIS) . /FTId=VAR\_009779."  
711  
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Region                    /note="Q -> E (IN PAIS) . /FTId=VAR\_013476."  
712  
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Region                    /note="L -> F (IN PAIS) . /FTId=VAR\_009780."  
715  
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/note="V -> M (IN PROSTATE CANCER; GAIN IN FUNCTION) .  
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Region                    717  
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/note="K -> E (IN PROSTATE CANCER) . /FTId=VAR\_009782."  
720  
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/note="K -> E (IN PROSTATE CANCER; FOUND IN BONE  
METASTASES) . /FTId=VAR\_009783."  
Region                    721  
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Region                    722  
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723  
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Region                    724  
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Region                    725  
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Region                    726  
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727  
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728  
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Region                    730  
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Region                    /note="V -> M (IN PROSTATE CANCER) . /FTId=VAR\_004695 ."  
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Region                    /note="D -> N (IN CAIS) . /FTId=VAR\_004696."  
732  
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Region                    /note="D -> Y (IN CAIS) . /FTId=VAR\_004697."  
733  
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Region                    /note="Q -> H (IN PAIS) . /FTId=VAR\_009792."  
737  
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741  
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742  
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742  
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743  
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Region                    /note="G -> E (IN CAIS) . /FTId=VAR\_013477."  
743  
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Region                    /note="G -> V (IN PAIS AND CAIS) . /FTId=VAR\_004699."  
744  
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745  
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746  
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748  
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757 /gene="AR"  
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759 /gene="AR"  
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Region

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762  
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/FTId=VAR\_004704."  
763  
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OF ANDROGEN BINDING). /FTId=VAR\_004705."  
Region  
763  
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764  
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Region  
765  
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Region  
765  
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766  
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Region  
767  
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768  
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Region  
771  
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772  
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Region  
772  
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774  
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MUTATION). /FTId=VAR\_004709."  
Region  
774  
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ANDROGEN BINDING). /FTId=VAR\_004708."  
Region  
779  
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Region  
780  
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Region  
782  
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Region  
784  
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TRANSACTIVATION). /FTId=VAR\_004712."  
Region  
787  
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Region  
788  
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Region  
790  
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Region  
791  
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Region  
793  
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Region  
794  
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Region  
798  
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/FTId=VAR\_004715."  
Region  
806  
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Region  
807  
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/FTId=VAR\_004716."  
Region  
807  
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807  
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/FTId=VAR\_004717."  
Region                    810  
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812  
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Region                    814  
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820  
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821  
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827  
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830  
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Region                    831  
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831  
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/FTId=VAR\_004720."  
Region                    834  
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/FTId=VAR\_009832."  
840  
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Region                    840  
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840  
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Region  
840  
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Region  
841  
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Region  
842  
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846  
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Region  
854  
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Region  
855  
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855  
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Region  
856  
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Region  
863  
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Region  
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481 gytrppqgla gqesdftapd vwypggmvsr vpypsptcvk semgpwmdsy sgpygdmrle  
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901 vqvpkilsgk vkpiyfhtq

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